26 January 2018  DÉJERINE 100 YEARS THERAPIES ACROSS ONE CENTURY OF NEUROLOGY

2.30 PM  Introduction
Saba Motta - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

2.40 PM  Presentation
Giuseppe Lauria - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

2.50 PM  Monsieur Dejerine de la Salpêtrière
Fabio Simonetti - Fondazione IRCCS Istituto Nazionale dei Tumori, Milano

3.20 PM  Early onset hereditary neuropathies: natural history and trial readiness
Isabella Moroni - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

3.40 PM  The Next-Generation Sequencing revolution and its impact on an old disease.
Franco Taroni - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

4.00 PM  Inherited neuropathies: the pathway to treatment, where are we?
Davide Pareyson - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

4.20 PM  Discussion

Dr. Simonetti will make an historical overview of Jules Déjerine (1849-1917), successor of Fulgence Raymond to the chair of the Salpêtrière. His name is linked to the sporadic form of olive-ponto-cerebellar atrophy, facio-scapulo-humeral myopathy, hypertrophic neuritis and thalamic syndrome. He definitively delivers to the memory of neurologists "Sémiologie des affections du Système Nerveux", the first text in which neurological semeiotics is treated as a discipline, with formidable iconography and case series.

Dr. Moroni will provide an overview on hereditary peripheral neuropathies, and will focalize mainly on infantile onset forms. These are rare disorders presenting with heterogeneous clinical phenotypes and causing variable degrees of impairment and disability since infancy, when they can be associated with central nervous system involvement. The inheritance pattern can be autosomal dominant, recessive and X-linked. Compared with adults, children present with relatively more frequent AR or sporadic forms, raising challenges for the differential diagnosis with acquired neuropathies.

Dr. Taroni will give an overview of the molecular bases of Charcot-Marie-Tooth disease and related neuropathies. Given the great number of genes involved and the phenotypic overlap, diagnosis of these forms is challenging. The talk will illustrate how the Next-Generation Sequencing technology contributes to the field, also generating new insights and discoveries.

Dr. Pareyson will present the progresses of drug therapies for Charcot-Marie-Tooth disease (CMT) and related neuropathies. Particularly, for the most common forms of CMT, numerous promising compounds are under study in cellular and animal models, mainly targeting either the protein degradation pathway or the protein overexpression. Efforts are also devoted to develop responsive outcome measures and biomarkers for this overall slowly progressive disorder, with quantitative muscle MRI resulting the most sensitive-to-change measure.
Fabio Simonetti, Neurologist, works as consultant at Fondazione IRCCS Istituto Nazionale dei Tumori of Milan. As a scholar of the history of semiotics and neurology, by visiting university libraries in cities inside and outside of Europe, he has acquired materials that have allowed him to write a volume dedicated to eponyms in Neurology with Prof. Faustino Savoldi from the University of Pavia. He was an organizer and speaker for history seminars dedicated to neurology, collected in various publications.

Isabella Moroni works as Associate in Child Neurology, at Fondazione IRCCS Istituto Neurologico C. Besta, Milan. She is involved in clinical research for several genetic and acquired diseases of central and peripheral nervous system (neuropathies, myopathies, mitochondrial and metabolic encephalopathies, leukodystrophies). The main areas of research include: selection of patients with specific phenotypes to address for extensive genetic studies and define genotype-phenotype correlations; identification of new genetic forms; definition of natural history for rare hereditary disorders; elaboration and standardization of disability and quality of life specific scales for children with peripheral neuropathies; cooperation in observational and interventional trials.

Franco Taroni, Neurologist and Geneticist, heads the Unit of Clinical Pathology and Medical Genetics and the Unit of Genetics of Neurodegenerative and Metabolic Disease at the Carlo Besta Neurological Institute. His research interests are focussed on the molecular bases of several hereditary neurological disorders including spinocerebellar degenerations and peripheral neuropathies.

Davide Pareyson, Neurologist, is currently Head of the Functional Department of Rare Neurological Diseases and Chief of the Rare Neurodegenerative and Neurometabolic Diseases Unit, Fondazione IRCCS Istituto Neuromedico C. Besta, Milan. He has been working on the development of outcome measures for hereditary neuropathies and other rare diseases and has coordinated and participated in clinical trials and natural history studies in inherited and acquired neuropathies.
INCONTRI DI AGGIORNAMENTO
IN NEUROSCIENZE
Gennaio – Dicembre 2018
14.30 – 16.00

KNOWLEDGE IS SPEAKING, WISDOM IS LISTENING
Jimi Hendrix